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ABSTRACT

This information fact sheet on screening newborns to prevent mental retardation defines newborn screening and outlines how screening is performed. It discusses the six most common disorders resulting in mental retardation for which states most commonly screen. These include phenylketonuria, congenital hypothyroidism, galactosemia, maple syrup urine disease, homocystinuria, and biotinidase deficiency. A chart lists, for each of the 50 states and the District of Columbia, the status of screening for each of these disorders. (Contains 10 references.) (JDD)

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Newborn Screening To Prevent Mental Retardation

The Arc
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The Arc

Q&A

a national organization on mental retardation

NEWBORN SCREENING TO PREVENT MENTAL RETARDATION

What is newborn screening?

Newborn screening identifies biochemical or other inherited conditions that may produce mental retardation, other disabilities and/or death. Babies are screened for these conditions during the newborn period. These conditions are identified using tests on blood collected from a heel stick onto filter paper.

Over 300 gene disorders can be traced to specific biochemical defects referred to as inborn errors of metabolism, and many of these disorders result in mental retardation. A specific intervention is possible that may prevent brain damage in about one-third of the identified gene disorders. However, because many disorders are extremely rare and screening all newborns is expensive, mass screening is currently being justified for only a few disorders (Baumeister et al., 1993).

For what conditions do states screen?

In the United States, all states and the District of Columbia screen for phenylketonuria (PKU) and congenital hypothyroidism. Forty-four states and D.C. screen for galactosemia, while fewer than half screen for maple syrup urine disease, homocystinuria and biotinidase deficiency. In addition, 42 states and D.C. screen for sickle cell disease, and some screen for a few other rare disorders. The chart features the six most common disorders resulting in mental retardation for which states most commonly screen.

Phenylketonuria (PKU). PKU is an inherited condition affecting one in 12,000 newborns (Thoene, 1992). It prevents the affected infant from metabolizing, or using, phenylalanine normally. Phenylalanine is one of the essential amino acids found in all protein foods. Unless the condition is detected and treatment is initiated soon after birth, phenylalanine builds up in the blood and body tissues and is toxic to the brain, usually resulting in severe mental retardation.

Beginning with the use of a routine blood screening procedure developed in the 1960s (Guthrie, 1992), all states now screen newborns for phenylalanine levels with the result that mental retardation from PKU has virtually been eliminated. Treatment requires eliminating foods naturally high in protein to remove phenylalanine from the diet within the first weeks of life and maintaining a protein-restricted diet through adolescence and perhaps throughout life. Though treated children have normal intelligence, some of them may have substantial learning difficulties (Levy, 1990).

With the effectiveness of the treatment for PKU, a new problem has arisen—maternal PKU. Risks as a result of maternal PKU occur when a woman with PKU becomes pregnant after having discontinued the low phenylalanine diet. Discontinuing the diet during childhood or on entering adulthood was common practice until young women with PKU began to have babies with mental retardation. The medical community then realized that if the mother is not on a low phenylalanine diet, the fetus will be exposed to toxic levels of phenylalanine that may result in severe mental retardation or death. Research suggests that the risk is greatly reduced if women with PKU reinstate their special diet for a period prior to conception and continue to monitor carefully their phenylalanine levels during pregnancy (Koch & de la Cruz, 1991).

Congenital hypothyroidism. Hypothyroidism results from an inadequate supply of the thyroid hormone, thyroxine. Approximately one out of every 4,000 newborn infants has hypothyroidism (Levy, 1990). Untreated, the child's growth will be stunted, and mental retardation will occur from lack of stimulation of the brain by the hormone thyroxine.

All states now screen for hypothyroidism using the same filter paper blood spots as for PKU screening. The condition is treated by initiating thyroid hormone replacement treatment as soon as possible after birth. Follow-up studies of treated children indicate that the hormone is an effective and cost-efficient method for preventing mental retardation, although the children may later exhibit learning difficulties and behavior problems (Rovet, Erlich & Sorbara, 1989).

Galactosemia. Galactosemia is a disorder in newborns of galactose metabolism affecting about one in 50,000 infants (Thoene, 1992). Galactose is the major sugar found in milk. It is normally converted to glucose, another simple sugar, but in galactosemia, the enzyme needed for this conversion is missing. Galactose accumulates in and damages the body's cells and organs. The damage can lead to jaundice, severe mental retardation, growth deficiency, overwhelming infection and death (Thoene, 1992).

Galactosemia is treated by excluding milk and milk products from the diet (Thoene, 1992). Children treated with this special diet usually show satisfactory general health and growth.

Maple Syrup Urine Disease. This is an extremely rare hereditary disease (1:200,000), characterized by an unusual maple syrup odor in the urine (Thoene, 1992). Symptoms appear several days after birth and also include poor feeding, lethargy and coma. Death within three months of birth is common. If the infant survives longer than a few months, mental retardation is apparent (Thoene, 1992).

Maple syrup urine disease is caused by abnormal metabolism of three amino acids and is treated by a complicated infant formula that must begin immediately following early diagnosis. It can be diagnosed through the same blood sample used to screen for PKU and galactosemia. Prompt treatment and careful follow-up in maintaining the special diet have helped many children to have normal intelligence.

Homocystinuria. This metabolic disorder results from a deficiency of the enzyme, cystathionine, and is estimated to occur in 1 in 200,000 newborns (Levy, 1990). Cystathionine is needed by the brain for normal development. Some of the symptoms of homocystinuria include dislocated lenses of the eyes, mental retardation, skeletal abnormalities and abnormal blood clotting.

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Twenty-one states now test newborns for homocystinuria. Homocystinuria is treated with a methionine-restricted diet supplemented with cystine. Early dietary treatment seems to prevent mental retardation and perhaps skeletal abnormalities. Dislocation of the ocular lenses occurs in many patients with homocystinuria, in spite of good dietary control (Levy, 1990). Other treatments are also being investigated (Thoene, 1992).

Biotinidase Deficiency. This is a disorder in which biotin, a member of the vitamin B complex, is deficient. Incidence is estimated at 1 in 70,000 live births (Baumeister et al., 1993). Symptoms include an abnormal red rash on the skin, poor muscle control, seizures, immune system impairment, hearing .oss and mental retardation. Usually symptoms are not noticeable at birth, but develop around three months of age.

Treatment for the disorder is very simple and inexpensive. As soon as the disorder is diagnosed, infants are provided with oral biotin supplements. With biotin treatment, symptoms of the disorder disappear. Left undiagnosed and untreated, it can result in coma and death. It is diagnosed with a urine test (Thoene, 1992).

National Newborn Screening Status Report

	Phenylketonuria	Congenital Hypothyroidism	Galactosemia	Maple Syrup Urine Disease	Homocystinuria	Biotinidase
Alabama	•	•	•			
Alaska	•	•	•	•	•	•
Arizona	•	•	•	•	•	•
Arkansas	•	•				
California	•	•	•			
Colorado	•	•	•	•	•	•
Connecticut	•	•	•	•	•	•
Delaware	•	•	•	•	•	•
D.C.	•	•	•	•	•	
Florida	•	•	•			
Georgia	•	•	•	•	•	
Hawaii	•	•				
Idaho	•	•	•	•	•	•
Illinois	•	•	•			•
Indiana	•	•	•	•	•	
Iowa	•	•	•	•		
Kansas	•	•	•			
Kentucky	•	•	•			
Louisiana	•	•				
Maine	•	•	•	•	•	
Maryland	•	•	•	•	•	•
Massachusetts	•	•	•	•	•	
Michigan	•	•	•	•		•
Minnesota	•	•	•			
Mississippi	•	•	•			
Missouri	•	•	•			
Montana	•	•	•			
Nebraska	•	•			•	•
Nevada	•	•	•	•	•	•
New Hampshire	•	•	•	•	•	
New Jersey	•	•	•			
New Mexico	•	•	•			•
New York	•	•	•	•	•	
North Carolina	•	•	•			
North Dakota	•	•	•	•		
Ohio	•	•	•		•	
Oklahoma	•	•	•			
Oregon	•	•	•	•	•	•
Pennsylvania	•	•		•		
Rhode Island	•	•	•	•	•	
South Carolina	•	•	•			
South Dakota	•	•				
Tennessee	•	•	•			
Texas	•	•	•			
Utah	•	•	•			
Vermont	•	•	•	•	•	
Virginia	•	•	•	•	•	•
Washington	•	•				
West Virginia	•	•	•			
Wisconsin	•	•	•	•	•	
Wyoming	•	•	•	•	•	•
Total Count	51	51	44	25	23	16

How is screening performed?

For the blood screening procedure, a sample of blood from the newborn infant is captured on a piece of filter paper from a prick to the heel. The blood dries on the paper and is sent to a laboratory where several types of tests can be performed. This method of specimen collection and transport to the testing laboratory was first made popular with the use of the test for PKU developed by Dr. Robert Guthrie. Based on PKU screening success and automation of the procedure, tests for other disorders were developed that could be applied to the same filter paper specimen that was submitted for PKU screening.

While many programs in the 1950s utilized urine screening for detecting metabolic disorders, these programs quickly switched to blood testing following publication of Guthrie's methodology in 1963 (Guthrie & Susi, 1963). Today there are few, if any, newborn screening programs which utilize urine testing. On the other hand, some infant screening programs use urine collected a few months after birth to screen for disorders such as neuroblastoma (Lemieux, 1988). Parents obtain the specimen by pressing a filter paper card onto the baby's wet diaper. For such testing a specimen kit is usually sent home with the parents when the baby leaves the hospital. The specimen is then mailed to a laboratory for testing.

In order to handle newborn screening efficiently and cost-effectively in smaller states, regional centers have been developed with adequately equipped and staffed laboratories where tests are performed on newborn blood specimens from all states in the region. In most other states, a single centralized laboratory is utilized. This concept of centralized laboratory testing also leads to improved reliability of the testing procedure and was particularly important when screening for hypothyroidism was implemented. The more complex technology required for hypothyroid screening was much more sophisticated and required more expertise than did the relatively simple PKU test (Therrell, 1993).

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